



CCND2 gene

cyclin D2

Normal Function

The *CCND2* gene provides instructions for making a protein called cyclin D2. Cyclins are a family of proteins that control how cells proceed through the multi-step cycle of cell division. Cyclin D2 helps to regulate a step in the cycle called the G1-S transition, in which the cell moves from the G1 phase, when cell growth occurs, to the S phase, when the cell's DNA is copied (replicated) in preparation for cell division. Cyclin D2's role in the cell division cycle makes it a key controller of the rate of cell growth and division (proliferation) in the body.

The cyclin D2 protein is regulated by a chemical signaling pathway called the PI3K-AKT-mTOR pathway. This signaling influences many critical cell functions, including the creation (synthesis) of new proteins, cell proliferation, and the survival of cells. The PI3K-AKT-mTOR pathway is essential for the normal development of many parts of the body, including the brain.

Health Conditions Related to Genetic Changes

megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome

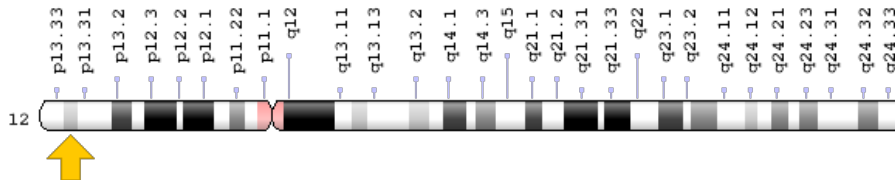
At least seven mutations in the *CCND2* gene have been found to cause megalencephaly-polymicrogyria-polydactyly-hydrocephalus (MPPH) syndrome. This rare condition affects the development of the brain, causing an unusually large brain and head size (megalencephaly) and a brain abnormality called bilateral perisylvian polymicrogyria (BPP). Some affected individuals also have an extra finger or toe on one or more of their hands or feet (polydactyly).

Each of the known mutations changes a single protein building block (amino acid) in the cyclin D2 protein. These changes prevent the protein from being broken down (degraded) when it is no longer needed. The resulting buildup of cyclin D2 in cells triggers them to continue dividing when they otherwise would not have, leading to abnormal cell proliferation. In the brain, the increased number of cells leads to rapid and abnormal brain growth starting before birth. It is less clear how a buildup of cyclin D2 contributes to polydactyly, although the extra digits are probably related to abnormal cell proliferation in the developing hands and feet.

Chromosomal Location

Cytogenetic Location: 12p13.32, which is the short (p) arm of chromosome 12 at position 13.32

Molecular Location: base pairs 4,273,736 to 4,305,356 on chromosome 12 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- G1/S-specific cyclin-D2
- KIAK0002
- MPPH3

Additional Information & Resources

Educational Resources

- Marie Curie Bioscience Database: PI-3K and AKT Signalling Pathway
<https://www.ncbi.nlm.nih.gov/books/NBK5964/#A40945>
- Molecular Biology of the Cell (fourth edition, 2002): Components of the Cell Cycle Control System
<https://www.ncbi.nlm.nih.gov/books/NBK26824/>

GeneReviews

- MPPH Syndrome
<https://www.ncbi.nlm.nih.gov/books/NBK396098>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28CCND2%5BTI%5D%29+OR+%28cyclin+D2%5BTI%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

OMIM

- CYCLIN D2
<http://omim.org/entry/123833>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_CCND2.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=CCND2%5Bgene%5D>
- HGNC Gene Family: Cyclins
<http://www.genenames.org/cgi-bin/genefamilies/set/473>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=1583
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/894>
- UniProt
<http://www.uniprot.org/uniprot/P30279>

Sources for This Summary

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<http://omim.org/entry/123833>
- GeneReview: MPPH Syndrome
<https://www.ncbi.nlm.nih.gov/books/NBK396098>
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